CASE 1994-8

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Reference number: ME 93-0357

Clinical History: This patient was well until age 42, when she was first noted to have a change in personality. Over the next two years, she developed a progressive dementia that resulted in an inability to function in her job as a secretary and eventually to be limited to the point of even having difficulty shopping. In her early thirties, there had been a transient episode of numbness and tingling in her left upper extremity, but she had no other history of medical problems. There was no prior history of hypertension.

When she was 44 years old, she was admitted to New York Hospital for neurologic evaluation. At that time, she was normotensive and general physical examination was normal. Neurologic examination revealed moderate cognitive impairment without aphasia. There was diffuse symmetric hyperreflexia, bilateral Hoffman's signs and bilateral Babinski signs with normal strength and gait.

CT scan revealed "white matter degeneration". Two years later, MRI of the brain revealed diffuse increase in signal on T2-weighted images throughout the cerebral hemispheric subcortical white matter. CBC, electrolytes, general chemistry panel, VDRL, B12, folate, thyroid function tests were normal. "Metabolic studies" were normal. CSF protein, glucose and cell count were normal.

She experienced further decline in neurologic function and was placed in a nursing home at the age of 48 years, when she was no longer able to ambulate. She died of pneumonia at age 56.

Autopsy findings: Gross autopsy findings revealed bilateral acute bronchopneumonia, mild systemic atherosclerosis and nephrolithiasis. External examination of the 1061 gram brain revealed bifrontoparietal and ventral brainstem atrophy. Internal examination revealed diminution in amount and gray discoloration of cerebral subcortical white matter, with multifocal cavitation, sparing U-fibers and polar regions. The basal ganglia, thalamic nuclei and pontine base had multiple small cavitary lesions.

Material submitted: One color 2x2 slide and one H&E section of cerebrum.
Points for discussion: 1. Diagnosis 2. Pathogenesis 3. Prognosis for family